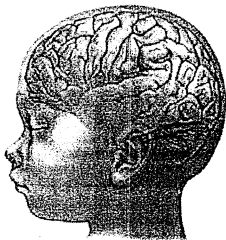
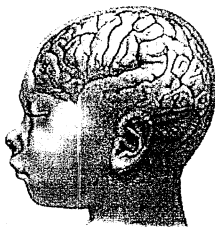


Neurology

Normal head size



Microcephaly



Hydrocephalus

In hydrocephalus the volume of CSF is increased within ventricular or sub-arachnoids spaces $\Rightarrow$ dilatation of the ventricles + $\uparrow\uparrow$ intracranial pressure in the majority of cases.

CSF- Circulation

$\Rightarrow$ next paper.

Etiology

A. Obstruction of CSF circulation (non-communicating)

i. At foramen of monoro.

- a. Congenital.
- b. Tumors.
- c. Inflammatory.

ii. At aqueduct of sylvius:

1. Congenital:

- Atresia.
- Stenosis.
- Forking.
- Vein of Galen.

2. Acquired:

- Gliosis.
- Tumors.

III. At the foramen of Magendi & Lushka.

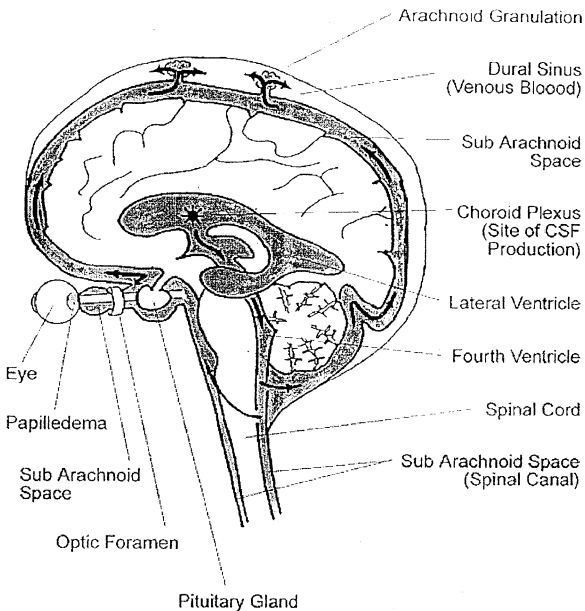
1. Congenital:- Dandy- Walker malformation.
2. Acquired:

- Meningitis.
- Hemorrhage.
- Posterior fosse tumors

B. Non obstructive (communicating):

i. Increased secretion:

1. Choroiditis.
2. Choroid congestion.
3. Pseudo tumor cerebri ..See later.
4. Meningism.



II. Decreased absorption:

1. Subarachnoid hemorrhage.
2. Subarachnoid adhesions.
3. Sinus thrombosis.
4. Achondroplasia.
5. Arnold Chiari syndrome.
6. Mucopolysaccharidosis.

TYPES



A. Obstructive: There is an obstruction to the CSF circulation between ventricles or at outlets of the 4th ventricle

B. Communicating: There is free communication between the ventricles & subarachnoid space i.e. ↑↑ production or ↓↓ absorption.

Arnold Chiari Malformation

- There is downward displacement of medulla oblongata & cerebellum ⇒ Obstruction of the subarachnoid pathways around the brain stem
- It is usually associated with spina bifida + Meningo - myelocele.

Clinical Picture

A. Before Closure of fontanels

1. ↑↑↑ Skull circumference.
2. Sutures ⇒ widely separated.
3. Fontanels ⇒ widely opened.
4. The scalp veins ⇒ dilated & visible.
5. The skin ⇒ stretched, shiny & thin.
6. Eyes ⇒ sun set appearance ⇒ why? & Optic atrophy in long standing cases.
7. The cry ⇒ high pitched.
8. Mac Ewen's sign ⇒ resonant note upon percussion of the skull, due to separation of the sutures.
9. Craniotabes.
10. Marked occipital expansion due to dilatation of the 4th ventricles in Dandy-Walker malformation.
11. Trans-illumination of the skull may be +ve in severe cases.
12. Neurological manifestations:
 - MR ⇒ due to atrophy of the cerebral cortex.
 - Convulsions.
 - Motor retardation & UMNL.



B. After closure of fontanels

The cause is usually acquired.

1. Signs of ↑↑ intra-cranial pressure (ICP).
e.g. persistent vomiting, severe headache & papilloedema
Disturbed tone & reflexes in the form of spasticity & hyperreflexia.
2. Skull is not significantly enlarged.
3. Mentality may be less affected.

Investigations

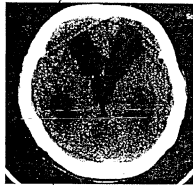
1. Plain X-ray skull

- Cranio-facial disproportion.
- Opened sutures & fontanels
- Intra-cranial calcification may be present (scattered in toxoplasmosis & peri-ventricular in CMV infection).
- Evidence of calcification in a tumor or hematoma may be present.
- Occipital bone expansion in Dandy- Walker malformation.

2. CT-Scan & MRI:

Showing $\Rightarrow$

- Size of ventricles.
- Site of obstruction.
- Pathology of obstruction.
- Extent of the disease.
- Remaining of cerebral mantle.



3. U/S of the brain: When the anterior fontanel is still opened.
4. Fundus examination: May show chorio-retinitis in congenital infection.
5. Serology: For STORCH infection.
6. Ventricular CSF pressure monitoring: Is the only accurate method to assess the activity of hydrocephalus "progressive or stationary"
7. Positron emission topography (PET):
8. Spectroscopic PET



I. From other causes of $\uparrow\uparrow$ skull circumference:

1. Rickets.
2. Achondroplasia.
3. Chronic hemolytic anemia.
4. Hydranencephaly.
5. Megalencephaly.
6. Familial.

II. From other causes of $\uparrow\uparrow$ ICP:

1. Space occupying lesion.
2. CNS infection.
3. Meningism.
4. Pseudotumor cerebri.
5. Encephalopathy.
6. Cranio Stenosis.
7. Severe hypertension.
8. Vasculitis syndromes.

Treatment

A. Observation & evaluation

1. If the patient is clinically well, with normal rate of skull growth $\Rightarrow$ continue observation.
2. In mild cases $\Rightarrow$ A trial of diuretics

B. Medical ttt (preoperative or if surgery is not indicated):

1. Diuretics e.g.
 - a. Diamox "Carbonic anhydrase inhibitor"
50 - 75 mg / kg / D $\Rightarrow$ $\downarrow$ C.S.F. Production about 1/3
 - b. Mannitol 200 mg / Kg over 20 minutes.
2. Penicillin for congenital syphilis.
3. Daraprim "Pyrimethamine" $\Rightarrow$ Toxoplasmosis.
4. Salt & water restriction.
5. Dexamethasone.
6. Anticonvulsants if there is convulsions.

C. Surgical ttt

1. Surgical removal of the cause of obstruction if possible e.g. cyst, tumors, abscess & hematoma.
2. Shunting the CSF to by pass the obstruction:

This is done by using a one way valve Spitz-Holter valve" The CSF is shunted to another site to be absorbed e.g.

 - a. Ventriculo-atrial shunt $\Rightarrow$ to the Lt atrium.
 - b. Ventriculo-peritoneal shunt $\Rightarrow$ to the peritoneal cavity.
 - c. Ventriculo-pleural shunt.
 - d. Ventriculo-cisternal shunt.

Once a successful shunt has been established- it should be maintained for life.
3. Choroids plectectomy or diathermy coagulation $\Rightarrow$ for choroids papilloma.
4. Third ventriculostomy in obstruction of foramen of Monroe.

Complications of shunt

A. Obstruction

Due to slipping, kinking or thrombus formation $\Rightarrow$
Manifestations of ICP

B. Infection

$\Rightarrow$ manifestations of septicemia as fever, rigors & toxemia.

C. Pulmonary embolism.

D. Focal glomerulonephritis.

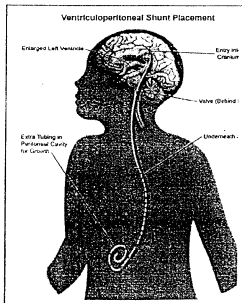
E. Chronic brain malfunctioning.

F. Shortening by age.

G. Acute shunt failure: in which there is increased ICP without evidence of obstruction or infection but symptoms improve on changing the shunt.

Causes of pseudo-tumor cerebri:

1. Hyper or hypovitaminosis A.
2. Hypoparathyroidism.
3. Hypervitaminosis D.
4. Outdated tetracycline.
5. Adolescent obese females.



Cerebral Palsy

Definition

- Non progressive central motor neuron deficit affecting the growing brain, dating to events in the prenatal, natal or postnatal period.
- Manifested since birth or early infancy.
- Prevalence 2-5 /1000 live births.
- More in LBW.



Hypertonia in C.P

Characters

1. Non fatal, non curable, non progressive.
2. Purely motor "no sensory manifestations"
3. May be associated with epilepsy, MR, deafness, speech, vision, behavioral & emotional disturbances.
4. Manifestations are present early in infancy or in the neonatal period.

Etiology

A. Ante- natal

1. Intra-uterine infections: STORCH.
2. Fetal anoxia $\Rightarrow$ Maternal hge
Placental insufficiency.
3. Maternal irradiations of the pelvis or drugs during pregnancy.
4. Congenital malformations of brain or vascular occlusion.
5. Some inborn errors of metabolism.

B. Natal

1. Birth injury $\Rightarrow$ IC hge.
2. Cerebral anoxia e.g. Hypoxic Ischemic Encephalopathy.

C. Post-natal

1. I.C. Infections: Meningitis, Encephalitis.
2. I.C. hemorrhage.
3. Neonatal asphyxia.
4. Bilirubin encephalopathy.
5. Metabolic disorders : Hypoglycemia,
hypocalcaemia & toxins.

Clinical Picture

A. Spastic C.P.(75 %)

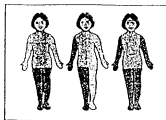
UMNL $\Rightarrow$

- Weakness or paralysis.
- Hypertonia more in the adductors $\Rightarrow$ scissoring of legs & antigravity Ms.
- Exaggerated deep tendon reflexes.
- +ve Babiniski sign
- Late muscle wasting.



• Distribution:

- Hemiplegia. *one side*
- Paraplegia. *both ll*
- Quadriplegia. *four limbs*
- Diplegia. *both sides of the upper*
- Monoplegia. *one limb*
- Double Hemiplegia. *one side of both upper*
- Biplegia. *upper of both*



MR $\Rightarrow$ Common finding.

The most common type is spastic diplegic C.P.

B. Atonic C.P.

- Severe hypotonia.
- Normal or exaggerated deep tendon reflexes.
- MR $\Rightarrow$ constant.
- High mortality.



C. Ataxic CP (1-2 %):-

$\Rightarrow$ Picture of cerebellar ataxia.

- Dysmetria.
- Tremors.
- Nystagmus.
- Zigzag gait.
- Dysidiadocokinesia.
- Hypotonia.



D. Extra-pyramidal CP (20 %):-

- Chorea.
- Athetosis.
- Dystonia.
- Static tremors.
- Abnormal tone $\Rightarrow$ hypotonia or rigidity.

E. Mixed C.P.

Functional & prognostic classification

According to the degree of motor dysfunction:

Class I: absent.

Class II: Mild.

Class III: Moderate.

Class IV: Severe.

Investigations

A. To reach the underlying etiology:

1. Plain X-ray skull.
2. Brain CT scan & MRI may be needed.
3. Fundus examination.
4. Lumbar puncture.
5. Metabolic screening as urinary plasma amino gram.
6. Serology for congenital infections.

B. For functional assessment:

1. I.Q.
2. Audiometry.
3. Psychometry
4. EEG

Treatment

1. Physiotherapy.
2. Speech therapy.
3. Special educational, social, psychological hygienic programs.
4. Nutritional rehabilitation.
5. Auditory aids.
6. Drugs which decrease the tone as baclofen, dantrolen Na, benzodiazepine, L- DOPA & alcohol
7. Anticonvulsants if there is epilepsy.
8. Surgery $\Rightarrow$ Orthopedic neurosurgery.

Abnormalities of size & shape of the skull

A. Microcranium.

B. Macrocephaly:

Macrocephaly

Definition

OFC $>$ 95th centile or OFC $>$ 2 SD for age, sex race.

Etiology

A. Cranial (thick skull bones):-

1. Chronic hemolytic anemia.
2. Rickets.
3. Osteogenesis imperfecta.
4. Osteopetrosis.
5. Hyperphosphatemia.
6. Familial.



B. Intracranial

1. Hydrocephalus.
2. Hydranencephaly replacement of the brain by CSF
3. Megalencephaly (increase in the number & size of the brain cells)
 - MPS.
 - MSUD.
 - Galactosemia.
 - Achondroplasia.
4. Porencephaly (hemispheric cyst)

Micro-cranium

Definition

It is small head with skull circumference less than the 3rd percentile

TYPES

A. Small brain ⇒ Micro-cephaly

B. Premature closure of the sutures ⇒ Cranio-synostosis "Cranio-Stenosis"

Microcephally

Etiology

1. Brain developmental defect:

- Primary (hereditary) AR micro-cephaly: It is characterized by backward sloping of the front.
- IEM ⇒ PKU MSUD.
- Fetal irradiation ⇒ 1st 2nd trimesters
- Fetal alcohol syndrome.
- Some chromosomal disorders as Down, Cornelia de Lange & Rubenstein Taybi syndromes.

2. Familial

3. 2ry to

- Congenital infections: STORCH-EB.
- Placental insufficiency & Peri-natal anoxia ⇒ Hypoxic ischemic encephalopathy.
- Post meningitis or post encephalitis.

Clinical Picture

1. Small skull.
2. No manifestations of ↑↑↑ ICP.
3. Early, fontanel is open.

Normal head size



Microcephaly



Investigations & Treatment

As C.P.

Cranio-Stenosis

Micro-cranium 2ry to premature closure of one or more of skull sutures.

CP

A. Abnormal shape of the skull

This depends on the affected sutures $\Rightarrow$

1. Sagittal suture: $\Rightarrow$ Dolico-cephaly or scapho-cephaly.
2. Bilateral coronal suture: $\Rightarrow$ Brachy-cephaly.
3. Unilateral closure of the coronal or lambdoid suture $\Rightarrow$ Plagio-cephaly.
4. Closure of all sutures: Oxycephaly (pointed head).
5. Metopic suture $\Rightarrow$ Trigono-cephaly triangular head with prominent vertical ridge in the mid-forehead.
6. Acrocephaly: $\Rightarrow$ Tower like head with vertical forehead.

C. Manifestations of $\uparrow\uparrow\uparrow$ ICP.

Investigation

X-ray skull shows $\Rightarrow$

- Silver beaten appearance.
- Abnormal shape of the skull.
- Bony closure of the suture.

Treatment

1. Craniotomy $\Rightarrow$ opening of the suture line.
2. Craniectomy $\Rightarrow$ removal of a bony strip it is very effective

Mental Retardation

Definition

It is a state of impairment of intelligence (complex mental process including thinking, memory, reasoning, verbal expression and adaptive behavior) dating from early life.

Etiology

A. Environmental

- No organic causes can be found.
- It due to socio-cultural & emotional disturbances.
- It is the most common cause of MR in about 90% of cases.
- It is usually mild.

B. Organic causes

1. Pre-natal natal

2. Chromosomal abnormalities

e.g. Trisomy 21, 18, 13 & deletion 5P - , 4P-

Fragile X- Chromosome

3. Post-natal ⇒ as CP.+

- Cerebral laceration due to head trauma.
- Infections as meningitis or encephalitis.
- Cerebro-vascular accident,
- Post-immunization encephalopathy e.g. Pertussis.
- Poisoning e.g. Carbon monoxide.
- Metabolic as severe hypoglycemia.

Degrees of MR

Degree	Term	IQ	Prognosis	Incidence
Mild	Moron	50-70	Educable	85%
Moderate	Imbecile	35-50	Trainable	5-10%
Severe	Idiot	20-35	Non-trainable 5%	
Profound	Idiot	0-20		

In some literatures moderate is referred to as high imbecile & severe as low imbecile.

How to calculate the IQ:

1. Assess the mental age by the developmental history. If level of different parameters are near to each others, consider the average.
2. If there is marked discrepancy the socio-adaptive history is the most important.
3.
$$IQ = \frac{\text{Mental age}}{\text{Chronological age}} \times 100$$

Manifestations of MR:

1. Delayed development of milestones & sphincter control.
2. Impaired learning ability.
3. Poor social adjustment.

Investigations & Treatment

As CP except no need for physiotherapy.

Hypotonia & neuromuscular diseases

Tone is the resistance of the muscle to stretch and the muscle is said to be hypotonic or flaccid when it offers little resistance to stretch.

Diseases affecting the anterior horn cells

- A. Poliomyelitis: see notebook of infection.
- B. Spinal muscular atrophy.
- C. Amyotrophic lateral sclerosis: very rare.
- D. Pena-Shokeir & Marden-Walker syndromes there is degeneration of motor neurons + arthrogryposis + multiple congenital anomalies.

Spinal muscular atrophy

Etiology

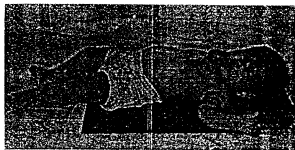
AR disease $\Rightarrow$ intrauterine degeneration of motor neurons.

Types

- 1. Infantile SMA (severe) $\Rightarrow$ Werdnig Hoffman.
- 2. Late infantile SMA 2 (mild).
- 3. Juvenile SMA3 (chronic).
- 4. Fazio-Londe $\Rightarrow$ bulbar more than spinal.

C/P of Werdnig Hoffman disease

- 1. Both sexes are affected.
- 2. +ve Family history + history of consanguinity.
- 3. History of weak fetal kicks.
- 4. After birth there is:
 - Weakness or paralysis.
 - Hypotonia.
 - Hypo-reflexia.
 - Later muscle wasting.
 - Distribution: All these findings are bilateral, symmetrical, proximal as distal.
 - Marked muscle fasciculations especially in the tongue.
 - Bulbar affection $\Rightarrow$ nasal tone of voice, nasal regurgitation of food, hoarseness of voice, dysphagia & repeated choking.
 - Staring look due to sparing of extra ocular & facial muscles.
 - Normal sphincters & intelligence.



Werdnig Hoffman disease

Investigations

1. EMG $\Rightarrow$ Neuropathic.
2. Nerve conduction velocity (NCV) $\Rightarrow$ normal.
3. Serum CPK: normal.
4. Muscle biopsy: picture of early denervation.

Treatment

Supportive tt because there is no available tt up till now. Some of later presenters may be trained to use computer.

Guillain Barre Syndrome

(Post- infections polyneuropathy)

Etiology

- Auto-immune post-infectious disease affecting the myelin sheath.
- It usually follows measles, chicken pox or influenza.

Clinical Picture

1. History of viral infection 1-3 weeks before the onset of the disease.
2. Acute motor weakness starting in the L.L. $\Rightarrow$ trunk $\Rightarrow$ UL $\Rightarrow$ bulbar "Landry's ascending paralysis"
3. Distribution: Usually bilateral, symmetrical, distal more than proximal of LMNL.
4. Tender calf ms.
5. Gloves & stoking hypothesia.
6. Others.
 - a. Bulbar palsy & other cranial Ns.
 - b. Respiratory affection.
 - c. Autonomic $\rightarrow$ Labile BP & HR
 - d. Encephalitis "Encephalo- myelo-radikulopathy cranialis" + papilloedema.
7. Chronic course may occur.

Investigations

1. EMG $\Rightarrow$ Neuropathic pattern.
2. Nerve conduction velocity $\Rightarrow$ Impaired.
3. CSF examination: cyto-albuminous dissociation
Oligo-clonal gammopathy.
4. Nerve biopsy: in chronic cases.

Treatment

Self limited in most cases

1. Steroid therapy.
2. IV immunoglobulin or prednisone pulse therapy.
3. Plasmapheresis.
4. Support of vital systems.
5. physiotherapy.

Other causes of polyneuropathy

1. Traumatic: Erb's palsy (neonatology), phrenic nerve palsy.
2. Peroneal muscular atrophy.
3. Refsum's disease.
4. Leukodystrophies.
5. Neurofibromatosis.
6. Toxic neuropathies: lead poisoning, arsenic, chemotherapeutics, INH.
7. Leprosy.
8. Vitamin B1, B6 & B12 deficiency.
9. Endocrinal as DM.
10. Familial dysautonomia.
11. Congenital insensitivity to pain.
12. Interstitial hypertrophic polyneuropathy.

Myasthenia Gravis

Types

1. **Congenital MG:** Due to congenital absence of acetyl choline receptors.
2. **Juvenile MG:** Autoimmune.
3. **Neonatal MG:** Infant of juvenile myasthenia mother.

Juvenile Myasthenia Gravis

Definition

A disease characterized by easy fatigability on repetition of muscular activity which is relieved by prostigmine.

Etiology

An auto-immune disease, where there are Antibodies directed against the motor end plate & compete with the action of Ac.ch.

CLINICAL

The disease affects the skeletal ms in a descending manner.

A. Ocular type

Ptosis, diplopia & squint by the end of the day.

B. Bulbar type

- Dropping of the jaw after prolonged chewing.
- Dysphagia & regurgitation of food from the nose.
- Dysarthria & nasal tone after prolonged talking.

C. Generalized type

- UL more involved than LL
- Proximal ms more than distal one.
- Respiratory muscles may be involved $\Rightarrow$ Dyspnea.

D. May be associated with:

- Thyrotoxicosis or hypothyroidism due to Hashimoto's thyroiditis.
- Thymic hyperplasia or adenoma.

E. Myasthenic crises:

Usually precipitated by infection, operation or idiopathic $\Rightarrow$ Severe respiratory distress

Diagnostic tests

1. **Induction of fatigue** $\Rightarrow$ by repetition of movement.
2. **Walker's test**
 - Apply sphygmomanometer cuff to one arm & raise above the systolic BP $\Rightarrow$ let the patient moves his fingers till fatigue.
 - When cuff is removed $\Rightarrow$ Ptosis occurs within 10 sec.
3. **Prostigmine "Neostigmine" test**
IM Prostigmine $\Rightarrow$ improvement after 15 mm.
4. **Edrophonium test**
IV Edrophonium $\Rightarrow$ improvement after 2 mm.

Investigations

1. EMG $\Rightarrow$ Reduction of amplitude on repetition of movement.
2. CXR $\Rightarrow$ May show Thymic shadow enlargement.
3. Thyroid profile
4. Anti Acetyl choline Antibodies.

treatment

A. Cholinesterase inhibitor drugs.

- Neostigmine.
 - 0.04mg / kg I.V
 - 0.4 mg / kg oral
- Pyridostigmine.
 - Toxicity $\Rightarrow$ cholinergic crises.

B. Steroid therapy

C. Plasma pheresis

Other causes of neuromuscular junction blockade

A. Botulism.

B. Organo-phosphorus compound poisoning.

D. Tic paralysis

- History of exposure to tic bite.
- Usually quadriplegia with shock stage.

Myopathies

Definition

These are diseases characterized by degeneration of muscle fibers & replacement by fibro-fatty tissues.

Etiology

I. Congenital:

1. Myo-tubular myopathy.
2. Nemaline myopathy.
3. Central core myopathy.
4. Mitochondrial myopathy.
5. Amyoplasia
6. Benign congenital hypotonia
7. Arthrogryposis multiplex congenita
8. Torticollis

II. Endocrinal & metabolic disorders:

1. Hypothyroidism (Kocher Debre Semi-laigne Syndrome).
2. Hyperthyroidism.
3. Hyperparathyroidism.
4. Steroid induced myopathy: Cushing syndrome.
5. Potassium related periodic paralysis.
6. Glycogen storage disease.
7. Vitamin E deficiency.

III. Inflammatory:

1. Dermatomyositis.
2. Polymyositis.
3. Viral myositis.
4. Parasitic myositis.

IV. Muscular dystrophy: characterized by 4 criteria.

- ◆ 1ry myopathy.
 - ◆ Genetic basis.
 - ◆ Progressive.
 - ◆ Muscle fiber degeneration & degeneration.
1. Duchenne.
 2. Becker's
 3. Facio-scapulo-humeral.
 4. Limb girdle myopathy.
 5. Ocular myopathy.
 6. Myotonia dystrophica.

Pseudo-hypertrophic Type (Duchenne myopathy)

Genetics

XLR "X P 21"

The gene is related to the Dystrophin protein of the sacrolemmal membrane. It is present in the muscles (skeletal & smooth), heart & brain, all of which are affected in the disease.

Patho-Physiology

- In myopathy the muscles are unable to convert creatine to creatinine to get energy.
- So $\Rightarrow$ there will be deficiency of energy $\Rightarrow$ degeneration of muscle fibers which are replaced by fibro-fatty tissue.
- According to the amount of fibro-fatty tissue we may get atrophy or pseudo-hypertrophy
- The creatine which is not converted excreted as such in urine.

G/P

Age of onset 5-15 years

A. Muscle weakness or paralysis of LMNL

- Bilateral, symmetrical affection of proximal ms.
- Hypotonia & hypo-reflexia.
- Winging of the scapulae $\Rightarrow$ "due to weakness of serratus anterior"
- Exaggerated lumbar lordosis & +ve Gower sign $\Rightarrow$ "due to weakness of extensors of the trunk"
- Pot belly abdomen $\Rightarrow$ "due to weakness of abdominal muscles"
- Waddling gait $\Rightarrow$ due to weakness of gluteus medius & minimus".
- +ve Slipping sign.
- Cardiomyopathy is nearly constant.
- Intellectual impairment is constant, but MR is present in only 20-30%
- Epilepsy more than in normal population.



lumbar lordosis, Pot belly abdomen

The following muscles never to be affected:

1. Sternomastoid muscles.
2. Upper fibers of trapezius muscle.
3. Clavicular head of pectoralis major

B. Pseudo — hypertrophy of some muscles.

- 1.L.L. $\Rightarrow$ calf & gluteus medius.
- 2.U.L. $\Rightarrow$ Triceps, Deltoid, Supra & infra-spinatus.

C. No sensory or sphincters affection.

D. Skeletal deformities.

- Pes cavus
- Kyphoscoliosis.

Causes of death

1. Pneumonias.
2. Intractable HF.
3. Sleep apnea.
4. Frequent aspiration.
5. UTI.

Investigations

1. EMG $\Rightarrow$ myopathic pattern .
2. Muscle biopsy $\Rightarrow$ replacement of the muscles by fibro-fatty tissues.
3. Enzymes $\Rightarrow$ $\uparrow\uparrow$ CPK, $\uparrow\uparrow$ Aldolase, $\uparrow\uparrow$ SGOT
4. Urine analysis $\Rightarrow$ $\uparrow\uparrow$ creatine & $\downarrow$ creatinine.
5. Pre-natal diagnosis $\Rightarrow$ by detection of the specific gene "XP21"
6. ECG & Echo-cardiography $\Rightarrow$ for cardiac affection.
7. Mothers of cases usually carriers & can be diagnosed by detection of slight increase of CPK or by genetic study.

Treatment

No hopeful ttt

1. Physio-therapy
2. Care of nutrition.
3. Recently $\Rightarrow$ Gene therapy & myoblast transplantation.
4. ttt of complications $\Rightarrow$ Pneumonia - HF - UTI.

Becker's Myopathy

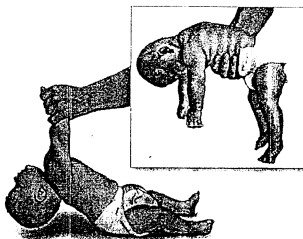
Very similar to Duchenne

Except:

1. More gradual course & delayed age of presentation.
2. Death is delayed to the late twenties.

Floppy baby

Floppy infant is an infant with severe persistent hypotonia present at birth or in early infancy.



Q1: How to diagnose floppy infant?

Diagnosis of severe hypotonia depends on the presence of the following signs:

1. **Frog-leg position**: In supine position, the limbs are abducted and slightly flexed simulating frog legs. This denotes hypotonia of limbs.
2. **Curved trunk on ventral suspension**: When the baby is suspended in prone position over the examiner's hand, he droops around it. This denotes hypotonia of the trunk muscles.
3. **Head lag**: When the baby is pulled up from his hands, while in supine position, the head lags backwards. This denotes hypotonia of neck muscles.
4. **Vertical suspension**: When hypotonic infant is suspended vertically, the head falls forwards, the legs dangle and the infant may slip through the examiner's hands because of weakness in shoulders.

Q2 : Is it central or neuromuscular?

Clinical differentiation between central and neuromuscular causes depends on deep tendon reflexes and mental development.

1. **Central causes:** characterized by:

- Normal or exaggerated reflexes.
- MR is common.
- Abnormal brain function e.g. marked decrease in level of consciousness & recurrent convulsions may be present.
- Dysmorphic features in chromosomal disorders.

2. **Neuromuscular causes:** are characterized by:

- Weak or absent tendon reflexes.
- Normal mentality.
- Muscle atrophy suggests motor neuron disease.
- Combination of atrophy & fasciculation is a strong event of denervation.
- No other organ malformation except joint & bone structures.

Causes of floppy baby

Central

1- CP

Atonic
Ataxic

2-Chromosomal

Cri-du-chait
Trisomy 13
Down

3- Others

Lowe\$
Pradder Willi

Peripheral

Spinal cord

Werdnig Hoffmann
Extensive polio
Meningo-myelocoele.

P.N.

G.B. \$
Post-diphtheritic
Lead poisoning.
Drug induced.
Axonal neuropathy.

MNJ

-M.G.
- Organophosphates toxicity.
- Botulism.
- Tick paralysis

MS

Myopathy:
Congenital.
Metabolic.
Inflammatory
Dystrophy

Causes of inability to walk

I. Non paralytic causes:-

1. Physiological delay.
2. Mental retardation e.g. Down syndrome, cretinism.
3. Bone diseases e.g. rickets.
4. Painful limb condition e.g. arthritis, osteomyelitis, Scurvy due to subperiosteal Hge.
5. Bad general condition e.g. malnutrition, chronic illness.
6. Congenital hip dislocation

II. Paralytic causes:-

1. Cerebral causes:-

- ♦ Cerebral palsy.
- ♦ Hydrocephalus.
- ♦ Post-meningitis or post-encephalitic sequellae.
- ♦ Post-traumatic sequellae.
- ♦ Post-hemorrhagic sequellae.

2. Spinal cord causes:-

- ♦ Poliomyelitis.
- ♦ WHD.
- ♦ Meningo myelocoele.
- ♦ Transverse myelitis.
- ♦ Pott's disease.
- ♦ Spinal cord trauma.

3. Peripheral neuritis:- as floppy baby.

4. Myopathies:-

- ♦ Congenital Myopathies.
- ♦ Inflammatory Myopathies.
- ♦ Muscular dystrophies.

Intracranial hypertension

It is the rise of ICP > 15 mmHg.

Causes:

1. Cranio synostosis.
2. Intracranial space occupying lesion.
3. Intracranial infection.
4. Meningism.
5. Vasculitis & hypertensive encephalopathy.
6. Pseudo-tumor cerebri..... see before.

It is syndrome of:-

- ♦ High ICP.
- ♦ Normal CSF content.
- ♦ Normal brain.
- ♦ Normal or small ventricles.

It is called benign as spontaneous recovery generally occurs, however serious visual complications may occur.

Diagnosed after exclusion of other causes of increased ICP.

Neurological complications of failure of extra-uterine adaptation

1. Hypoxic ischemic encephalopathy (HIE).

2. I.C He due to:

- ♦ Birth injuries.
- ♦ HIE.
- ♦ 1ry hemorrhagic disturbances.
- ♦ Congenital vascular anomalies.
- ♦ Fragile vessels as in premature.

3. CNS infections as meningitis due to :-

- ♦ Defective immunity.
- ♦ Different technical maneuvers in the neonatal ICU.

4. Bilirubin encephalopathy due to

- ♦ Excess unconjugated Bilirubin load.
- ♦ Defective blood brain barrier.
- ♦ Precipitating factors as hypoxia, acidosis, hypothermia or hypoglycemia.

5. Endocrinal abnormalities as congenital hypothyroidism.

SEIZURES

Terminology

- **Seizure** :paroxysmal cerebral dysrhythmia.
- **Convulsions** : motor seizure.
- **Fit**: one attack.
- **Epilepsy**: recurrent seizures unrelated constantly to fever or brain insult.
- **Tonic convulsions**: continuous muscle contractions.
- **Clonic**: repetitive contraction and relaxation.
- **Myo- Clonic**: jerky like movements which are stereotypic but not eventually rhythmic.

Etiology of seizures

1. **Congenital brain abnormalities** as Microcephaly, lessencephaly.
2. **Infections**
 - Meningitis.
 - Encephalitis.
 - Both.
 - Abscess.
 - Sinus thrombosis.
3. **Vascular**
 - Embolism.
 - Thrombosis.
 - Vasculitis.
4. **Tumors 1st or 2nd**.
5. **Head trauma** concussion or lacerations.
6. **Encephalopathy's**, e.g.
 - * Hepatic.
 - * Water intoxication.
 - * Renal.
 - * Lead.
 - * Hypertension.
 - * CO₂.
 - * Hypoxic.
 - * CO
7. **Metabolic**, e.g.
 - Hyponatremia.
 - ↓ Ca
 - ↓ Mg
 - ↓ glucose
 - B6 deficiency.
 - B6 dependency.
 - Inborn errors of metabolism.
8. **Other space occupying lesions** as: Cyst or tuberculoma.
9. **Neuro-degenerative diseases**.
10. **Idiopathic**.
11. Febrile convulsions.
12. **Drugs** as digoxin, amphetamine, and analeptics.

Clinical classification of epilepsy:

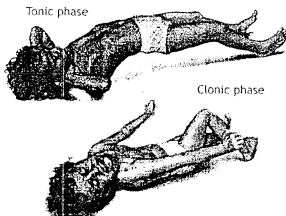
I. Generalized:

- Absence.
- Tonic.
- Myo-Clonic.
- Infantile spasms.
- Tonic Clonic.
- Clonic.
- Atonic

II. Partial.

- Simple
 - * Motor.
 - * Sensory
 - * Autonomic.
 - * Psychic.
- With secondary generalization.
- Complex.

III. Unclassified



(1) Simple partial:

(No loss of consciousness)

- Motor $\Rightarrow$
- Sensory $\Rightarrow$ Jacksonian fits.
- Psychic
- Autonomic $\Rightarrow$ abdominal epilepsy

(2) Complex partial (loss of consciousness):

aura $\Rightarrow$ automatism.

(3) Absence seizures:

A. Simple = Petit mal.

B. Complex = motor activities.

It might be associated rarely with falling.

(4) Generalized tonic Clonic seizures.

- $\Rightarrow$ $\pm$ aura.
- $\Rightarrow$ epileptic cry
- $\Rightarrow$ tonic $\Rightarrow$ seconds
- $\Rightarrow$ Clonic.

Febrile convulsions

It is fever-related convulsions which cause is not yet settled.

Criteria

- Age: 3m- 5 years.
- Neurologically free before and after
- EEG normal 2 weeks after.
- Very high fever > 38.8°C.
- Rapid rise of temp.
- Infection should not be central (extra-cranial).
- Attack lasts: seconds $\Rightarrow$ 10 mm (not >15mm).
- No recurrence in one fever (allowed once within 12 h).
- Recurrent seizure attacks can be up to 3 times (some say 5).
- Seizures are Clonic & generalized.

It may turn to epilepsy if the following risk factors were present:

1. +ve family history.
2. Onset < 9 m. age.
3. Prolonged or atypical.
4. Delayed development.
5. Abnormal neurological signs.

Treatment

- Control of fever
- Treatment of cause.
- Short term anticonvulsants.
- Avoid fever later.

DIAGNOSIS OF CONVULSIONS

Age Consideration

1. Neonatal
 - Trauma
 - Intra-cranial hemorrhage
 - Hypoxia
 - Metabolic
 - Congenital anomalies
2. 6 m—5 years —
 - Febrile convulsions
 - Breath holding attacks
 - Then other causes.
3. After 5 years
 - Idiopathic epilepsy
 - No febrile convulsions
 - Others.

Course of Convulsions

1. Acute non recurrent

- Febrile
- Trauma
- Infections
- Toxic
- Cerebral edema
- Metabolic
- Other encephalopathy's

2. Chronic recurrent

- Idiopathic epilepsy
- Tumors
- Neurodegenerative.
- Epilepsy like states---
- Hysteria
- Narcolepsy
- Cataplexy
- Migraine

Associated Features:

1. Signs of meningitis.
2. Type of seizures.
3. Neurological abnormalities
4. Mental retardation.

Investigations of seizures:

1. 1.EEG False +ve
± Prolonged EEG monitoring with simultaneous closed circuit video recording.
± Brain mapping.
2. CT or MRI if focal
3. Angiography or Doppler.
4. CSF study.
5. Biochemical studies.
6. Fundus examination.

Treatment

Is it seizure?

- (I) No = paroxysmal non epileptic
1. Benign paroxysmal vertigo.
 2. Breath holding.
 3. Cough syncope.
 4. Familial choreo-athetosis
 5. Narcolepsy.
 6. Night tremors.
 7. Rage attacks.

(II) Yes ⇒

i. First attack ⇒ EEG, CT and CSF (usually febrile and metabolic)

1. Abnormal ⇒ treatment of the cause

- Anti epileptic drugs in acute phase.

- Observation.

2. All normal ⇒ Observation.

3. Normal except EEG ⇒ Classify seizure type.

ii. Recurrent search for a cause and classify seizure type into either good responders or poor ones.

1. Maintain cardiopulmonary function.

2. Anticonvulsant drugs in acute stage status epilepticus.

a) Diazepam or Lorazepam IV or rectal. then

b) Phenytoin 15 - 20 mg/kg IV short very slowly 1 mg/kg/mm, then
5- 8 mg/kg/day divided on two doses.

Some centers use phenobarbitone before phenytoin if diazepam was ineffective.

c) Diazepam drip or Paraldehyde or General anesthesia.

3. After control ⇒ follow up drugs, see table

4. ACTH usually in infantile spasm.

5. Ketogenic diet.

6. Surgery.

b) If viral was suspected.

- * Virus isolation e.g.
 - CSF $\Rightarrow$ Herpes simplex hominis, rubella, enterovirus.
- * Serology $\Rightarrow$ 1gM (= recent fetal source).
 $\Rightarrow$ IgG (maternal source).

3. Routine:

a) CBC:

- * $\uparrow$ or $\downarrow$ TLC or Normal
- * $\downarrow$ Platelets
- * $\uparrow$ ESR
- * Band cells.
- * Shift to left.

b) $\uparrow$ C-reactive protein.

c) Liver, renal functions.

4. Imaging:

- a) X-ray chest (even without pulmonary symptoms).
- b) X-ray long bones.
- c) X-ray skull calcified (Cytomegalovirus & toxoplasma)
- d) Ultra-sonography for the abdomen and skull.

Treatment

I. Antibiotics:

- * Immediate.
- * Aggressive (doses calculated according to age;).
- * According to culture & sensitivity or broad spectrum combination.
- * Bactericidal.
- * Take your samples before treatment.
- * Best combinations are:
 - Penicillin + amino glycosides.
 - Penicillin + 3rd generation cephalosporins.
- * Better IV. or even drip.
- * Continue the course 2 weeks or at least 5-7 days after response
- * If no response in 2 days:
 - Change the drugs or think of pus collection.

II. Supportive treatment:

- * Fluid and electrolytes (take care of SIADH).
- * Blood product therapy.
- * Treatment of D.I.C.
- * Exchange transfusions or granulocyte transfusion.
- * Assisted ventilation.
- * IV immunoglobulins.
- * Fibronectin therapy.

Antibiotics according to their capability to pass blood brain barrier and their safety in the newborn:

1. 3rd generation cephalosporins
2. Penicillin.
 - * Non inflamed meninges (do not pass).
 - * inflamed meninges (pass).

NEONATAL MENINGITIS

Definition

Infection of the lepto-meninges of the newborn infants.

Etiology

1. common: (Early < 1 week).

E.coli & group B streptococcus are most common.

2. Less common (late > 1 week):

- * Staph. Aureus
- * Klebsiella
- * Pseudomonas
- * Proteus
- * Listeria monocytogenes

Route:

1. Early onset:

- ⇒ Amniotic fluid ⇒ severe form
- ⇒ birth canal ⇒ severe form

2. Late onset: ⇒ from community.

Clinical Picture:

- Usually non-specific, so that the need for scoring system was essential (sepsis score).
Local features are rare if ever.
- General:
 - Not doing well.
 - Weak suckling
 - Vomiting
 - Sclerema neonatorum.
 - Poor control of temperature
 - Lethargy.
 - Sluggish neonatal reflexes.
- Local:
 - Irritability
 - Hypertonia
 - Hypo or hyperreflexia
 - Abnormal neonatal reflexes.
 - Hypotonia
 - Tremors or seizures.
 - Bulging fontanel.

Complications:

- * DIC
- * Shock
- * Heart failure
- * NEC
- * Cardiac arrest
- * Syndrome of inappropriate anti-diuretic hormone secretion.
- * Cerebral palsy, MR or epilepsy later if survived.

Diagnosis

1. Maternal history: e.g. - Prenatal rupture of membrane.

2. Identification of the causative organisms:

- * Culture ⇒ bacteria
- * Serology ⇒ virus.
- a) If bacteria was suspected ⇒
 - * Gram stained smear of different specimens; blood, CSF, etc.
 - * Blood, CSF ⇒ culture and sensitivity.
 - * Counter immune-electrophoresis ⇒ rapid detection of bacterial agents.